



## SGCA gene

sarcoglycan alpha

### Normal Function

The SGCA gene provides instructions for making the alpha component (subunit) of a group of proteins called the sarcoglycan protein complex. The sarcoglycan protein complex is located in the membrane surrounding muscle cells. It helps maintain the structure of muscle tissue by attaching (binding) to and stabilizing the dystrophin complex, which is made up of proteins called dystrophins and dystroglycans. The large dystrophin complex strengthens muscle fibers and protects them from injury as muscles tense (contract) and relax. It acts as an anchor, connecting each muscle cell's structural framework (cytoskeleton) with the lattice of proteins and other molecules outside the cell (extracellular matrix).

### Health Conditions Related to Genetic Changes

#### limb-girdle muscular dystrophy

More than 70 mutations in the SGCA gene have been identified in people with limb-girdle muscular dystrophy type 2D. Limb-girdle muscular dystrophy is a group of related disorders characterized by muscle weakness and wasting, particularly in the shoulders, hips, and limbs. Forms of limb-girdle muscular dystrophy caused by gene mutations that affect the sarcoglycan complex are called sarcoglycanopathies.

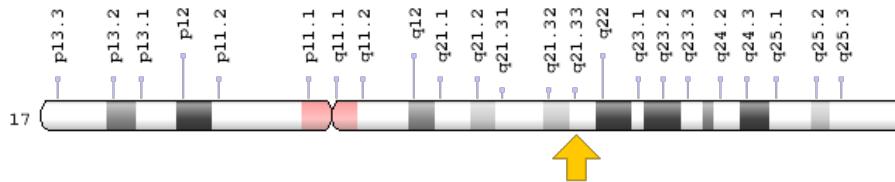
The most common SGCA gene mutation occurs in about one-third of people with limb-girdle muscular dystrophy type 2D. This mutation replaces the protein building block (amino acid) arginine with the amino acid cysteine at position 77 in the alpha-sarcoglycan protein, written as Arg77Cys or R77C. The rest of the known SGCA gene mutations are specific to individual families or certain populations.

SGCA gene mutations may prevent the sarcoglycan complex from forming or from binding to and stabilizing the dystrophin complex. Problems with these complexes reduce the strength and resilience of muscle fibers and result in the signs and symptoms of limb-girdle muscular dystrophy.

## Chromosomal Location

Cytogenetic Location: 17q21.33, which is the long (q) arm of chromosome 17 at position 21.33

Molecular Location: base pairs 50,165,517 to 50,175,932 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 50-DAG
- 50 kDa dystrophin-associated glycoprotein
- 50DAG
- 50kD DAG
- adhalin
- ADL
- alpha-sarcoglycan
- alpha-SG
- DAG2
- DMDA2
- dystroglycan-2
- LGMD2D
- sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein)
- SCARMD1
- SG-alpha
- SGCA\_HUMAN

## **Additional Information & Resources**

### Educational Resources

- University of Washington Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/musdist/lg.html#ad>

### GeneReviews

- Limb-Girdle Muscular Dystrophy Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1408>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SGCA%5BTIAB%5D%29+OR+%28sarcoglycan,+alpha%5BTIAB%5D%29%29+OR+%28%2850+kDa+dystrophin-associated+glycoprotein%5BTIAB%5D%29+OR+%2850-DAG%5BTIAB%5D%29+OR+%2850DAG%5BTIAB%5D%29+OR+%28adhalin%5BTIAB%5D%29+OR+%28alpha-sarcoglycan%5BTIAB%5D%29+OR+%28alpha-SG%5BTIAB%5D%29+OR+%28DAG2%5BTIAB%5D%29+OR+%28LGMD2D%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

### OMIM

- SARCOPHYCAN, ALPHA  
<http://omim.org/entry/600119>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SGCA.html](http://atlasgeneticsoncology.org/Genes/GC_SGCA.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SGCA%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=10805](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10805)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6442>
- UniProt  
<http://www.uniprot.org/uniprot/Q16586>

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